

Difficulties in the Diagnostics and Treatment of Near-Total Congenital Megacolon

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Rezumat

Dificultăți de diagnostic și tratament în megacolonul congenital - forma aproape completă

Forma aproape completă de aganglionoză intestinală este rar întâlnită în cadrul pacienților diagnosticați cu boală Hirschsprung și până în urmă cu câțiva ani era considerată fatală. Stabilirea diagnosticului corect s-a dovedit a fi dificilă, întrucât semnele clinice și radiologice nu sunt patognomonice. Obstrucția intestinală cronică și dependența de nutriție parenterală totală pe termen lung se asociază cu o rată crescută a mortalității în cazul acestor pacienți. Cu toate că nu există un consens cu privire la metoda ideală pentru cura chirurgicală a acestei patologii, pacienții beneficiază de tehnici precum coborârea intestinului cu inervație normală sau chiar transplantul de intestin. Vă prezentăm cazul unui nou-născut de sex feminin care a fost transferat în clinica noastră pentru distensie abdominală și vărsături biliocoloide, cu suspiciune de malformație de tub digestiv, la care diagnosticul de aganglionoză formă aproape completă s-a stabilit cu dificultate. S-a intervenit chirurgical, practicându-se intervenții seriate și alegându-se ca tehnică de alungire a intestinului metoda miotomie-miectomie extinsă (descrisă de Ziegler), cu evoluția postoperatorie imediată favorabilă. Pacienta dezvoltă însă un episod septic și, în ciuda tratamentului medicamentos

conform antibiogrammei, starea ei se deteriorează, decesul înregistrându-se la 77 de zile de la momentul internării.

Cuvinte cheie: Hirschsprung, aganglionoză totală, miotomie-miectomie, nutriție parenterală totală

Abstract

Near total colonic aganglionosis is one of the rarest forms of Hirschsprung's disease and until recent years it has been considered deadly. Establishing a correct diagnosis has proven to be challenging, because while the clinical and radiological features can be useful, they are not pathognomonic. Chronic intestinal obstruction and long-term parenteral nutrition dependency are associated with a high mortality risk for these patients. While there is no current consensus with regards to a superior operative method, the patients benefit from surgical techniques aimed at lengthening the intestine, as well as from intestine transplant. We report the case of a newborn baby girl who was admitted to our clinic for abdominal distension, biliary and fecaloid vomiting. With an initial suspicion of digestive tract malformation, the diagnosis of near total congenital megacolon was established with great difficulty and the infant underwent serial surgeries, ending up with an extended myotomy-myectomy (Ziegler's procedure) as a curative approach, with favorable immediate postoperative evolution. However, the patient developed sepsis and although the infection was treated accordingly, the baby's general condition kept deteriorating and exitus was recorded 77 days after admission.

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Key words: Hirschsprung, total aganglionosis, myotomy-myectomy, total parenteral nutrition

Introduction

Congenital Hirschsprung's disease (HD) is characterized by the absence of ganglion cells in the distal bowel. While the internal anal sphincter is the constant inferior limit, HD can be classified as classical HD - when the aganglionic segment does not extend beyond the upper sigmoid, long-segment HD - when the aganglionic segment extends to the splenic flexure or transverse colon and total colonic aganglionosis - when the aganglionic segment extends to the colon and a short segment of terminal ileum (1).

Total or near-total colonic aganglionosis (NTCA) represent the most extreme and rare form of congenital Hirschsprung's disease (2), being reported in only 3-12% of all HD cases (3). Although the incidence of HD is estimated to be 1 in 5000 births, with a male: female ratio of 4:1, it has been found that in the case of total or near-total colonic aganglionosis the ratio is reversed, with a female preponderance (1).

The diagnosis of near-total aganglionosis is difficult compared to other forms of HD and often causes delays in initiating the treatment (4). Because of this, patients can undergo multiple operations and frequently experience peri-operative complications (5). NTCA is considered to be a fatal condition in infancy because chronic intestinal obstruction and dependence to long-term parenteral nutrition are associated with high mortality and morbidity rates (2).

While there are several methods for the surgical management of NTCA, the difficulty surgeons are faced with is preserving an ideal intestinal length. It is well known that at least 40 cm of small intestine with normal innervation are required in order to ensure the absorption of vital nutrients. Extended myectomy-myotomy of the proximal jejunum (Ziegler's procedure) has been successfully used in order to achieve a functioning length of the intestine (6,7).

We report the case of a newborn baby girl who was transferred to the Surgical Department of our clinic for abdominal distension accompanied by bilious vomiting. After 3 surgical interventions, the pathological examination of the resected intestine established the diagnosis of NTCA. Two months after admission, the decision was taken to perform an extended myotomy-myectomy, with favorable immediate postoperative evolution. However, the patient developed sepsis and although the infection was treated accordingly, the baby's general condition kept deteriorating and exitus was recorded 77 days after admission.

Case report

We report the case of a newborn baby girl weighing 3100g,

who was transferred to our clinic for abdominal distension and bilious vomiting, with an onset at 36 hours after birth. On admission, the patient's general condition was poor, with dry skin, jaundice, persistent abdominal skin fold, bilio-fecaloid gastric aspirate, distended abdomen with generalized bloating and visible intestinal loop outline, with a small quantity of mucus spontaneously eliminated through the anus. A nasogastric tube was placed and antibiotic treatment was initiated.

The abdominal ultrasound performed on admission showed distended intestinal loops, with high peristalsis and the abdominal radiography showed normal shape and structure of the stomach, the pylorus, with early emptying, visible duodenal frame and dilated jejunal loops, intense abdominal pneumatisation, gas and fluid levels.

The patient underwent exploratory laparotomy, with intra-operative findings showing dilated ileal and jejunal loops, with semi-liquid content, the last ileal loops and the colon being smaller in volume, not dilated, full of meconium, with areas of calcification. A manual evacuation of the meconium was performed and the diagnosis of meconium ileus was established. Postoperatively, the patient's evolution was not favorable, with the development of an occlusive episode. On the 10th day after surgery, a second intervention was required and an ileostomy was placed. Because the patient's condition continued to deteriorate, with no stool being eliminated through the ileostomy, 10 days after the 2nd surgery, a third intervention was performed and a jejunostomy was placed at about 30 cm from the duodenojejunal flexure. The ileostomy was closed and serial biopsies of the ileum and colon were taken. The histopathological examination of the bioptic fragments revealed no autonomic nerve throughout the colon and the ileum, with rare ganglion cells found in the fragments taken from the jejunum. Based on these findings, the diagnosis of NTCA was established.

*Postoperatively, the patient's evolution was favorable, with stool passing through the jejunostoma. The patient received total parenteral nutrition; her condition improved and she gained weight. At the age of 2 months, the decision was taken to perform an extended myotomy-myectomy (Ziegler's procedure), with immediate favorable evolution. 15 days after the elongation procedure, the patient's general condition deteriorated and a reintervention was performed; perforation at the site of the myotomy was found and sutured. The patient then developed an episode of sepsis with double etiology, positive for *Klebsiella Pneumoniae* and *Candida*. Although the infection was treated accordingly, the baby's condition kept worsening and exitus was recorded 77 days after admission to our clinic.*

Discussion

Total or near-total colonic aganglionosis is characterized by the absence of ganglionic innervation throughout the entire or nearly entire gastrointestinal tract (2). This form of megacolon affects 3-12% of all infants with HD (4), with a slight female preponderance (male:female ratio is 0.8:1) (1). Patient's age at the moment of diagnosis has progressively decreased over the

years, by the age of 3 months, the diagnostic rate being almost 40% (1). The newborn with HD is usually a full-term baby and presents with a distended abdomen, bilious vomiting and delay in the passage of meconium (1,3).

Our patient was born at 38 weeks' gestation and, 36 hours after birth presented abdominal distension, bilious and then fecaloid vomiting, with a small amount of mucus being spontaneously eliminated through the anus.

The diagnosis of HD is supported by anamnestic data [families with a higher incidence of the disease (8)], clinical (early symptoms) and laboratory (barium enema, manometry) examinations and it is confirmed by the histopathological examination of the bioptic fragments. The diagnosis of near-total aganglionosis is difficult compared to other forms of HD and often causes delays in initiating the appropriate treatment (4). A common problem encountered in cases with NTCA is the failure to consider the diagnosis which leads to inappropriate procedures, the most common one being the placement of a stoma in the aganglionic intestine (5).

In our case, after the first surgical intervention, the diagnosis of meconium ileus was established and during the second surgery, an ileostoma was placed (aganglionic territory).

Although multiple procedures exist for the treatment of NTCA, there is no current consensus on a superior operative procedure. The treatment of NTCA involves early decompression of the colon by an ostomy, using the most distal ganglionated bowel. This is usually followed by a second reconstructive procedure, after the child has had adequate time to grow and nutritional abnormalities have been corrected (9). Early surgical procedures for the treatment of total and near-total colonic aganglionosis have evolved from the procedures used for other forms of HD, such as the pull-through procedures described by Swenson, Duhamel and Soave. In order to decrease morbidity rates, several modifications have been made to these procedures. Martin et al used the absorptive capacity of the remaining aganglionic colon through the latero-lateral anastomosis of the terminal ileum with the descending and sigmoid colon. This procedure was then further modified to use the entire colon. However, increasing the anastomotic length between the functional small bowel and the aganglionated colon has shown no improvement of the morbidity rates. Kimura, Boley and others have described the ascending colon patch technique (9).

The main problem surgeons are faced with is that resection does not leave enough normal bowel in order to support full/enteral feeding, prolonged parenteral nutrition being necessary. The need to increase the intestinal absorption surface has led to new surgical options being considered, which preserve a part of the aganglionic intestine, such as Ziegler's procedure, bowel tapering, STEP (serial tapering enteroplasty) and intestinal transplantation. Even so, most patients are dependent on total parenteral nutrition (TPN) and present high risk for TPN or disease-related complications in infancy (2,7).

In cases where the remaining ganglionic bowel length is less than 20 to 40 cm, the probability of irreversible intestinal failure is high and complications related to long-term

parenteral nutrition occur repeatedly. Intestinal transplantation seems to be the logical alternative treatment for such patients (3,10).

In our case, the timing of the therapeutic intervention was chosen taking into account the patient's general condition, which was improving, the elimination of stools through the jejunostoma, the absence of infectious episodes. An extended myotomy-myectomy was chosen, taking into account the length of the remaining bowel (30 cm) and estimating that the aganglionic intestine will act as a passive conduct able to receive and absorb nutrients propelled from the proximal intestine.

Total or near-total colonic involvement has a higher morbidity and mortality than short segment HD and requires a more complex pre- and postoperative management. NTCA has fatal outcomes because of sepsis caused by bacterial translocation or central venous line infection, liver failure determined by prolonged parenteral nutrition. Most patients die before reaching their second birthday (6).

While at first our patient's postoperative evolution was favorable, 15 days after the elongation procedure, her condition started deteriorating. She developed sepsis, with dual etiology, positive for *Candida* and *Klebsiella Pneumoniae*. In spite of correct antibiotic, antifungal and supportive treatment, the patient died 77 days after admission.

Despite the unfortunate outcome, we consider this case as having been a beneficial experience to the team of doctors involved in the care of a patient diagnosed with an extremely rare form of Hirschsprung's disease.

Conclusions

NTCA is a rare form of Hirschsprung's disease that was considered fatal in infancy. Improvements in supportive care, early and accurate diagnosis and appropriate treatment have led to an increased survival rate in these patients.

The diagnosis of this pathology is known to be difficult, because while the clinical and radiological features can be useful, they are not pathognomonic. There is no current consensus with regards to a superior operative method with respect to perioperative morbidity and mortality; the patients benefit from surgical techniques aimed at lengthening the intestine, as well as from intestine transplant. An important part in the management of NTCA is to prevent major complications due to prolonged dependence to total parenteral nutrition such as septic complications related to the central venous catheter, liver failure.

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